

11 β -hydroxylase deficiency: Twenty years follow-up

Ayla Güven, Didem Helvacioğlu, Suna Kılınc

Göztepe Training and Research Hospital, Pediatric Endocrinology Clinic, Istanbul, Turkey

Background:

We presented twenty-year follow-up of two patients with 11 β -hydroxylase deficiency, one of them has novel mutation.

Case Presentation:

Case 1: The male patient was diagnosed at the age of 9-months. He presented with penil growth and pubic hair. At diagnosis blood pressure was normal, his hormonal profile was distinctive of 11 β -OHD, with elevated serum levels of 11-deoxycortisol (DOC 134 ng/mL, N: 0-1.18). Hydrocortisone was introduced immediately after diagnosis. Analysis of the CYP11B1 gene revealed a novel missense mutations (R374G). At the age of 6, his testes were 4-5cc, bone age was 12 years. Precocious puberty was diagnosed also. He was treated with GnRH analogue and ciproteron acetat for the following 5 years. At age of 11 yr, he was hypertensive (140/100 mm Hg), antihypertensive therapy was started. At 19-years-old scrotal ultrasonography was consisted with adrenal rest tumor. His final height was not well within the normal range (162 cm). Currently, he is mildly overweight (body mass index, 26.2 kg/m²) and normotensive (100/70 mm Hg), and he receives glucocorticoid and antihypertensive therapy.

Case 2: The girl presented at age 2-months with external virilization. Growth acceleration was remarkable during childhood. Blood pressure was within the normal range. Basal DOC was elevated (8.5ng/ml). Analysis of the CYP11B1 gene revealed two previously known missense mutations (R43Q, A386V). Her height was 141.8 cm (+2.83 SDS) with an advanced BA of 16 yr when she was 8 yr. She had her menarche at 12 yr. Now she is 20-years old, her height is 142.1 cm (- 3.5 SDS) and menstrual periods are irregular

Conclusion:

11 β -hydroxylase deficiency though rare, to understanding the clinical symptoms as well as genetic analysis for early diagnosis and management of complications are important. We want to emphasize the complications of 11 β -OHD during long-term follow-up.

